Reviewer’s report

Title: Genetic analysis of 62 Chinese families with Duchenne muscular dystrophy and strategies of prenatal diagnosis in a single center

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Reviewer: Luisa-Maria Botella

Reviewer's report:

The manuscript is interesting and sound. This work is showing a large genetic analysis of DMD combining both, MLPA and NGS. The data supplied are rich and at the same time the design for the genetic analysis of families with genetic history of the disease is appropriate and sound. These data should be published.

Some concerns affect to difficulties in understanding certain paragraphs, and in the confusion between terms "de novo DMD mutations and new mutations". The authors should be aware of the following points:

1. In the abstract page 2 line 37 the word was should be included between study and aimed and the sentence reformulated as: "This retrospective study was aimed at supplying information on our 4-year clinical genetic and....................."

2. The sentence in line 48 page 2 is difficult to understand: in 52 families with positive results, 45 mothers showed the same mutations as the probands? How could it be possible, that the proband had a different mutation from the mothers, this fact should be commented in results or discussion since it seems striking

3. At the end of page 7, it is concluded that other molecular methods are requiered for those probands in 10 families with no positive findings of MLPA and NGS. This reviewer is assuming that the family has an history of DMD and therefore an index case, except if the "putative" index case is dead and there are no other cases in the family. Another possibility could be a mutation raised as "de novo mutation" in the embryos. In any case these explanations should be included.

Again, in page 8 lines 165 and 167 (table 1) 7 mothers out of 52 probands did not carry any mutation. For sure this are "de novo mutations appeared in the foetus, or in the first divisions of the zygote. If this is a possible explanation, please include it in the text.

4. In the discussion section, line 182 the sentence "next generation and strongly hope to determine" should be changed to "next generation and strongly wished to determine....."

5. Discussion, first paragraph page 10. It should be clear a difference or distinction between the terms "de novo" mutation, which means a mutation appearing in the proband and not inherited, from "new mutations, unpublished so far in literature or in the databases". This difference is crucial and we believe that the novel mutations referred to in the first paragraph of page 10 are new mutations, not reported so far in literature.

Figure 2 showing the changes in the sequence of the new mutations, should be composed in such a way
that in addition to the sequence alignment, the point of the capillary chromatography, where the nucleotide change appear as well as the surrounding sequence is shown in a Sanger chromatogram. This way is more illustrative and easy to follow.

In general a survey of English and editing of typos should be performed.

**Are the methods appropriate and well described?**
If not, please specify what is required in your comments to the authors.

Yes

**Does the work include the necessary controls?**
If not, please specify which controls are required in your comments to the authors.

Yes

**Are the conclusions drawn adequately supported by the data shown?**
If not, please explain in your comments to the authors.

Yes

**Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?**
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

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